Providing a cure for sphingosine phosphate lyase insufficiency syndrome (SPLIS) through adeno-associated viral mediated SGPL1 gene therapy

Grant Award Details

Providing a cure for sphingosine phosphate lyase insufficiency syndrome (SPLIS) through adeno-associated viral mediated SGPL1 gene therapy

Grant Type: Quest - Discovery Stage Research Projects

Grant Number: DISC2-13072

Investigator:

Name:	Julie Saba
Institution:	University of California, San Francisco
Type:	PI

Award Value: $1,463,400

Status: Pre-Active

Grant Application Details

Application Title: Providing a cure for sphingosine phosphate lyase insufficiency syndrome (SPLIS) through adeno-associated viral mediated SGPL1 gene therapy

Public Abstract:

Research Objective

AAV-SPL 2.0 is a gene therapy cure for SPLIS, a lethal childhood disorder of metabolism that causes kidney failure. Our gene therapy may also work in more common fibrotic (scarring) kidney diseases.

Impact

Our treatment may cure a rare but often fatal genetic disease (SPLIS) for which no specific treatment is available. It may additionally cure other forms of kidney disease caused by kidney scarring.

Major Proposed Activities

- Test the ability of our gene therapy to prolong survival in a newborn mouse model of SPLIS.
- Test the ability of our gene therapy to protect the kidney from damage in an adult mouse model of SPLIS.
- Test the ability of our gene therapy to protect the kidney from damage in mouse models of more common forms of kidney fibrosis.
- Use mouse models to demonstrate where in the body our gene therapy can reach and restore the activity of the enzyme encoded by the gene.
- Test the ability of the gene therapy to restore sphingolipid metabolism in mouse models of SPLIS

Statement of Benefit to California:

AAV-SPL 2.0 gene therapy may cure children with SPL insufficiency syndrome (SPLIS) and individuals with kidney disease arising from many common conditions and that can lead to chronic kidney disease and kidney failure. Patients with SPLIS have been diagnosed in California. Chronic kidney disease affects 3% of Californians, with higher rates in areas of agricultural work. It is more common in 65 and older adults and more common in Black and Hispanic adults compared with white and Asian adults.
